20200805_data_release_notes

Overview of submitted records: 2020

Jan 01, 2020	1,026,969
Feb 01, 2020	1,041,077
Mar 01, 2020	1,055,499
Apr 02, 2020	1,084,731
May 01, 2020	1,136,163
Jun 01, 2020	1,142,645
Jul 01, 2020	1,255,451
Aug 01, 2020	1,302,013

Overview of changes in the release of August 5, 2020

Content

Brief	Explanation
American College of Medical Genetics and Genomics (ACMG)	American College of Medical Genetics and Genomics (ACMG) submitted interpretations based on a practice guideline for 4 variants in FMR1 and Fragile X syndrome, and for 3 variants in DMPK and myotonic dystrophy 1.
ClinGen Platelet Disorders Variant Curation Expert Panel	ClinGen Platelet Disorders Variant Curation Expert Panel submitted 115 novel variant interpretations.
ClinGen PAH Variant Curation Expert Panel	ClinGen PAH Variant Curation Expert Panel submitted 89 novel variant interpretations.
ClinGen RASopathy Variant Curation Expert Panel	ClinGen RASopathy Variant Curation Expert Panel submitted 13 novel variant interpretations and updates to 2 records.
ClinGen Lysosomal Storage Disorder Variant Curation Expert Panel	ClinGen Lysosomal Storage Disorder Variant Curation Expert Panel submitted 82 novel variant interpretations.
Invitae	Invitae submitted 49,636 novel variant interpreations and updates to 45,278 records.
CeGaT Praxis fuer Humangenetik Tuebingen	CeGaT Praxis fuer Humangenetik Tuebingen submitted 760 novel variant interpretations and updates to 84,321.
Wong Mito Lab, Molecular and Human Genetics, Baylor College of Medicine	Wong Mito Lab, Molecular and Human Genetics, Baylor College of Medicine submitted 352 novel variant interpretations.
Foulkes Cancer Genetics LDI, Lady Davis Institute for Medical Research	Foulkes Cancer Genetics LDI, Lady Davis Institute for Medical Research submitted 330 novel variant interpretations.
Integrated Genetics /Laboratory Corporation of America	Integrated Genetics/Laboratory Corporation of America submitted 234 novel variant interpretations and updates to 13 records.
Laboratory of Genetics in Ophthalmology, Institut Imagine	Laboratory of Genetics in Ophthalmology, Institut Imagine submitted 214 novel variant interpretations.

Centogene AG - the Rare Disease Company	Centogene AG - the Rare Disease Company submitted 177 novel variant interpretations.
Johns Hopkins Genomics, Johns Hopkins University	Johns Hopkins Genomics, Johns Hopkins University submitted 149 novel variant interpretations.
Constitutional Genetics Lab, Leon Berard Cancer Center	Constitutional Genetics Lab, Leon Berard Cancer Center submitted 140 novel variant interpretations.
Elsea Laboratory, Baylor College of Medicine	Elsea Laboratory, Baylor College of Medicine submitted 127 novel variant interpretations.
NEI Ophthalmic Genomics Laboratory, National Institutes of Health	NEI Ophthalmic Genomics Laboratory, National Institutes of Health submitted 112 novel variant interpretations.
dbSNP build 154	We have updated ClinVar data with rs numbers from dbSNP build 154. Also rs numbers that were submitted are now reported in the SCV section of our XML products as a database cross-reference using the XRef element under the Measure, rather than with the Attribute element under Measure.
changes to single-letter protein changes	In this release, we changed the way we report single-letter protein changes, e.g. V600E: • we removed ~4000 values that we calculated previously but that are no longer supported by our processing • we stopped reporting single-letter codes for stop lost • we started reporting values that we did not calculate previously for missense variants that are not SNVs (e.g. for short insertions, deletions, or inversions)
coming soon: changes to variant position, reference and alternate alleles in variant_summary.txt	Soon we will update the way that the variant position and reference and alternate alleles are reported in variant_summary.txt. We will add columns for vcf_pos, vcf_ref, and vcf_alt. Variants that are in scope for our VCF file will have their position and ref /alt alleles reported in this way. Variants that are not in scope for our VCF file will have their position reported in the old fields: Start, Stop, ReferenceAllele, AlternateAllele. A variant is expected to be reported one way or the other, but not both. This change will make variant_summary.txt more consistent with ClinVar's VCV XML file. Please contact us as clinvar@ncbi.nlm.nih.gov if you have questions or comments about this change.
coming soon: change in how p. expressions are stored in XML	Soon we will change the way we handle p. expressions that are incomplete because they lack an accession.version, e.g. p. Tyr1863Cys. We will store these descriptions in the XML files as alternate names, rather than as HGVS expressions.

Overview of submitted records: 2019

Jan 01, 2019	759562
Feb 07, 2019	778673
Mar 01, 2019	782638
Apr 01, 2019	787656
May 01, 2019	795045
Jun 01, 2019	811551
Jul 01, 2019	819827
Aug 01, 2019	825177
Sept 01, 2019	881419
Oct 01, 2019	888298
Nov 01, 2019	889968
Dec 01, 2019	893196

Overview of submitted records: 2018

Jan 01, 2018	579543
Feb 01, 2018	582113
Mar 01, 2018	593651
Apr 01, 2018	610005
May 01, 2018	645149
Jun 01, 2018	676018
Jul 01, 2018	676575
Aug 01, 2018	685942
Sep 01, 2018	701880
Oct 01, 2018	708726
Nov 01, 2018	715516
Dec 01, 2018	749203

Overview of submitted records: 2017

Jan 01, 2017	396005
Feb 01, 2017	405182
Mar 01, 2017	406220
Apr 01, 2017	446265
May 01, 2017	482941
Jun 01, 2017	486420
Jul 01, 2017	488658
Aug 01, 2017	492592
Sep 01, 2017	504299
Oct 01, 2017	512373
Nov 01, 2017	517157
Dec 01, 2017	519359

Overview of submitted records: 2016

Jan 01, 2016	172867
Feb 01, 2016	176710
Mar 01, 2016	178032
Apr 01, 2016	180549
May 01, 2016	181155
Jun 01, 2016	192617
Jul 01, 2016	204415
Aug 01, 2016	209842
Sep 01, 2016	210200
Oct 01, 2016	213499

Nov 01, 2016	236420
Dec 01, 2016	240042

Overview of submitted records: 2015

Jan 01, 2015	149013
Feb 01, 2015	156999
Mar 01, 2015	162455
Apr 01, 2015	171408
May 01, 2015	172044
Jun 01, 2015	173236
Jul 01, 2015	184506
Aug 01, 2015	154686
Sep 01, 2015	158580
Oct 01, 2015	160538
Nov 01, 2015	170931
Dec 01, 2015	172006

Overview of submitted records: 2014

Jan 01, 2014	68204
Feb 01, 2014	73492
Mar 01, 2014	83343
Apr 01, 2014	111501
May 01, 2014	112349
Jun 01, 2014	117209
Jul 01, 2014	127132
Aug 01, 2014	127557
Sep 1, 2014	143114
Oct 1, 2014	143601
Nov 1, 2014	144117
Dec 1, 2014	148008

Overview of submitted records: 2013

Apr 05, 2013	30333
May 01, 2013	30386
Jun 01, 2013	39047
Jul 01, 2013	39170
Aug 01, 2013	45901
Sep 01, 2013	50263

Oct 01, 2013	52047
Nov 01, 2013	64750
Dec 01, 2013	64881